



Pseudoxanthoma Elasticum

Also known as ...

Grönblad–Strandberg Syndrome

What is Pseudoxanthoma Elasticum?

Pseudoxanthoma elasticum (PXE) is a progressive systemic disorder resulting from the accumulation of calcium and minerals in the elastic fibres of the connective tissue. Connective tissue provides strength and flexibility to your body structures. PXE can affect the elastic fibres in the skin, as well as the eyes, blood vessels and sometimes the digestive tract.

It affects approximately 1 in 50,000 people worldwide.

What causes Pseudoxanthoma Elasticum?

Pseudoxanthoma elasticum is a genetic disorder caused by mutation in the ABCC6 gene. This gene is important for making proteins found in cells of the liver, kidneys, skin, stomach, blood vessels and eyes.

It is inherited in an autosomal recessive manner. This means a person must have received both copies of the mutated genes from their parents to be affected. Most people can carry a copy of the mutated gene but not have the condition.

What does Pseudoxanthoma Elasticum look like?

People with PXE often have small yellowish bumps (papules) on their skin that resemble skin of a “plucked chicken”. the skin can become lax and redundant in advanced stages. These changes are typically seen on the neck, armpits and inner surface of joints.

What other problems can occur with Pseudoxanthoma Elasticum?

The abnormal deposition of minerals in PXE can cause abnormalities in the:

- Eyes – abnormal vessel growth resulting in bleeding or scarring of the retina. This causes changes in colour vision, reduced or complete loss of vision.
- Heart & Blood vessels – narrowing of the arteries causes cramping and pain in arms and legs (claudication), stroke, renovascular hypertension, angina & heart attacks.
- Digestive tract. – bleeding
- Obstetric complications: increased risk of miscarriage in the first trimester, worsening of the skin & systemic complications during pregnancy.

How is Pseudoxanthoma Elasticum diagnosed?

The diagnosis can be made clinically. A skin biopsy may be needed from skin lesions. It is formally diagnosed via genetic testing. Imaging, eye exam & further testing may be needed to assess other organs involvement.

How is Pseudoxanthoma Elasticum treated?

No specific treatment for PXE exists. The most important aspect of management is to ensure that complications are prevented & dealt with by the appropriate specialist. Regular Surveillance for complications is often recommended.

Lifestyle precautions

- Avoiding contact sports to reduce the risk of bleeding & eye injury
- Avoiding aspirin and NSAIDs to reduce the risk of digest tract bleeding
- Stopping smoking

What is the likely outcome of Pseudoxanthoma Elasticum?

Despite the possibility of PXE to affect multiple organs, most affected individuals live a normal life span. Affected individuals should be aware of the potential complications and to look out for these symptoms.

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